

Table 1 : Comparison of diagnostic methods for SMA

N° tube	SSCP ⁽¹⁾	Our method (radioactive) ⁽³⁾					
		Probe 1 (exon 7)		Probe 2 (exon 8)		Exon 7	Exon 8
Exons ⁽²⁾	PSL/mm ²	R (%) ⁽⁴⁾	Exon 7	PSL/mm ²	R (%) ⁽⁴⁾		
Control (1)	ndel 7/ndel 8	42	0	ndel	22	0	ndel
Control (2)	ndel 7/ndel 8	41	0	ndel	23	0	ndel
Control (3)	ndel 7/ndel 8	43	0	ndel	22	0	ndel
Control (4)	ndel 7/ndel 8	41	0	ndel	21	0	ndel
Control (5)	ndel 7/ndel 8	42	0	ndel	23	0	ndel
SMA (6)	del 7/del 8	24	43	del	15	32	del
SMA (7)	del 7/del 8	14	67	del	08	64	del
SMA (8)	del 7/del 8	10	76	del	07	68	del
SMA (9)	del 7/del 8	26	38	del	06	73	del
SMA (10)	del 7/del 8	09	79	del	13	41	del
SMA (11)	del 7/del 8	27	36	del	12	45	del
SMA (12)	del 7/del 8	15	64	del	15	32	del
SMA (13)	del 7/del 8	13	69	del	14	36	del
SMA (14)	del 7/del 8	25	40	del	11	50	del
SMA (15)	del 7/del 8	20	52	del	09	59	del
SMA (16)	del 7/del 8	19	55	del	13	41	del
SMA (17)	del 7/del 8	12	71	del	14	36	del
SMA (18)	del 7/del 8	14	67	del	12	45	del

(1) single strand conformation polymorphism

(2) del : deleted ; ndel : non deleted

(3) The quantification of results obtained is performed by means of Bio-Imager and expressed as PSL/mm²

(4) R : difference = 1 - [(PSL/mm²Control - PSL/mm²SMA)/(PSL/mm²Control)]

The mean value of the control group is used for the calculation of R

Table 2 : Comparison of diagnostic methods for SMA

N° tube	SSCP ⁽¹⁾	Our method (ELISA)							
		Probe 1 (exon 7)			Probe 2 (exon 8)			Probe 3 (HUMEF1AB)	
		Exons ⁽²⁾	Optical density	R(%) ⁽³⁾	Exon 7	Optical density	R(%) ⁽³⁾	Exon 8	Optical density
Control (1)	ndel 7/ ndel 8	0.26	0		ndel	0.28	0	ndel	0.55
Control (2)	ndel 7/ ndel 8	0.28	0		ndel	0.29	0	ndel	0.54
Control (3)	ndel 7/ ndel 8	0.27	0		ndel	0.26	0	ndel	0.52
Control (4)	ndel 7/ ndel 8	0.26	0		ndel	0.28	0	ndel	0.51
Control (5)	ndel 7/ ndel 8	0.27	0		ndel	0.27	0	ndel	0.49
SMA (6)	del 7/ del 8	0.16	41		del	0.13	54	del	0.51
SMA (7)	del 7/ del 8	0.16	41		del	0.16	43	del	0.48
SMA (8)	del 7/ del 8	0.11	59		del	0.12	57	del	0.53
SMA (9)	del 7/ del 8	0.15	44		del	0.14	50	del	0.49
SMA (10)	del 7/ del 8	0.19	30		del	0.16	43	del	0.5
SMA (11)	del 7/ del 8	0.13	52		del	0.12	57	del	0.49
SMA (12)	del 7/ del 8	0.14	48		del	0.13	54	del	0.48
SMA (13)	del 7/ del 8	0.17	37		del	0.14	50	del	0.55
SMA (14)	del 7/ del 8	0.12	55		del	0.15	46	del	0.52
SMA (15)	del 7/ del 8	0.11	59		del	0.13	54	del	0.5
SMA (16)	del 7/ del 8	0.13	52		del	0.15	46	del	0.51
SMA (17)	del 7/ del 8	0.15	44		del	0.12	57	del	0.49
SMA (18)	del 7/ del 8	0.16	41		del	0.16	43	del	0.5

(1) single strand conformation polymorphism

(2) del : deleted ; ndel : non deleted

(3) R : difference = $1 - [(OD_{450} \text{Control} - OD_{450} \text{SMA}) / OD_{450} \text{Control}]$

The mean value of the control group is used for the calculation of R